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FIG. 1. Profile of the proposita revealing 'bird-headed' facies, lobeless ears, micrognathia, and pro-optosis.

## Familial Bird-headed Dwarfism (Seckel's Syndrome)

**Summary.** Low birth weight dwarfism with mental retardation, large eyes, a beaklike nose, narrow face, receding mandible, and dental anomalies are the specific features of 'bird-headed dwarf of Seckel'. The following case report presents details of a Seckel dwarf with familial occurrence of the trait and, thus supports an autosomal recessive mode of inheritance. In addition, the possible significance of dental alterations is noted.

### Case Report

The proposita is a 46-month-old negro who possessed the clinical syndrome of Seckel's bird-headed dwarfism; the familial occurrence of the trait in three sibs is also included.

The patient was born 2 May 1969 in Osceola, Arkansas. The child weighed 1308 g at birth and 40 weeks' gestation. To date the patient presented as severely retarded, and weighing 4578 g.

The proposita had microcephaly and oxycephaly, with pro-optosis of the eyes, nystagmus, and lobeless ears (Fig. 1). Marked micrognathia was noted as was a central posterior cleft of the palate (Fig. 2). The dentition revealed severe hypoplasia of enamel in the primary dentition; however primary second molars showed no

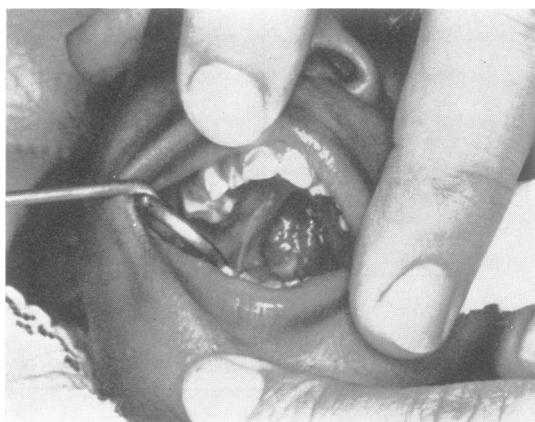


FIG. 2. Intra-oral view revealing posterior cleft palate and hypoplastic enamel defects of the primary dentition. The changes on the central incisor have been clinically modified for a restoration.

altered morphology and appeared to be radiographically normal with regard to morphology, location, and sequence of development. A hyperostotic ridge could be palpated along the sagittal sinus. The head was 30 cm in circumference.

Hip abduction was almost impossible and the extremities revealed scissoring and deep tendon reflexes with hyperactivity. The radiological examination noted congenital dislocation of the right hip, thinness of all long bones, and scoliosis of the thoracolumbar spine.

The skin was free of rashes or petechiae. The nose and throat were clear and benign except for a midline posterior cleft of the hard palate. The thyroid was not enlarged. The chest was symmetrical and shield-shaped with a measurement of 28 cm.

The heart was not enlarged and showed regular sinus

tachycardia. There were no thrills present. A murmur was present along the left sternal border and was best noted at the third and fourth intercostal spaces. Lungs were clear throughout both to percussion and auscultation. The abdomen was normal and the genitalia were infantile.

Haematology showed a haemoglobin of 10.2 g%, haematocrit of 34.5%, total red blood cell count of 4.19 million, white blood cell count 7750 with a differential of: 38% PMNL (seg), 5% PMNL (non-seg), 50% lymphocytes, 6% monocytes, 1% eosinophils; and a reticulocyte count of 0.67%. The platelet count was 456,000 cells/cm<sup>3</sup>. The MCV was 85, MCH 25, and MCC 29.5.

Immunochemistry revealed a serum IgG of 803 mg%, IgA 141 mg%, IgM 160 mg%, and salivary IgA 3.4 mg%.

Cytogenetics from peripheral leucocytes showed 46 chromosomes in each cell with a karyotypic analysis showing no deviation from the normal (46,XX).

Dermatoglyphics revealed a digital pattern where the distribution of ulnar loops were L1, L3, R1, R3, and R4. The distribution of whorls were L2, L4, L5, R2, R5. The *atd* angles were right 46° and left 40°.

The family history revealed that the patient was the product of a 27-year-old negro male and a 31-year-old, gravida 12 para 11, female negro. Both of the parents were in good health and revealed no congenital abnormalities. The proposita had seven sibs living, all of whom were in good health. One of the pregnancies ended in a miscarriage. Three infants were born alive and lived for 2, 3, and 5 months. These infants were reported to have features similar to the proposita. The respective birth weights of the microcephalics were 1308 g (proposita), 1580 g (first pregnancy), 1680 g (sixth pregnancy), and 1821 g (seventh pregnancy). The paternal grandmother was said to have 'some type' of congenital deafness. No other familial illnesses were known. Consanguinity was not apparent (Fig. 3).

Radiological examination of the skull and cephalometric indices were performed and compared with a paediatric standard (Coben, 1955). Although the standard represented a slightly older population it was

the youngest standard available and it was felt that it would reflect a trend towards the skeletal alterations that might exist if an ideal control was available.

From the radiological examination one could note extreme microcephaly with skull measurements of 7.62 cm lateral diameter, and 11.41 cm in an anterior-posterior diameter. The fontanelles appeared to be closed as were the sutures. The anterior cranial vault was arched upward. The sella tursica was normal.

Cephalometric analysis and comparison to a pediatric standard revealed that the angle ramal-plane to facial height was increased by five standard deviations over the control group. The mandibular-plane to facial height was two standard deviations greater than a control. The gonial angle and facial angle were both within normal limits. The convexity of the face was approximately 7 standard deviations greater than a control group (Table I).

TABLE I  
CEPHALOMETRIC INDICES COMPARED TO A  
PAEDIATRIC STANDARD

	Mean*	SD*	Range*	Seckel Dwarf
Ramal plane-facial height	9.8	4.98	-2/+19	39
Mandibular plane-facial height	26.4	4.07	18/36	39
Gonial angle	126.2	5.41	114/138	130
Facial angle	84.6	3.37	77/94	90
Convexity of the face	+4.8	4.14	-2/+15	34

\* Standard values for 8-year-old males and females from Coben (1955).

## Discussion

Seckel (1960) devoted a monograph to a group of patients for whom he used the designation 'bird-headed dwarf'. In that monograph he called attention to a specific type of dwarfism with mental retardation, low birth weight, small head, large eyes, a beaklike nose, narrow face, receding mandible, and dental anomalies. Warkany, Monroe, and Sutherland (1961) reported a similar case, with extreme microcephaly and posterior cleft palate. Harper, Orti, and Baker (1967) added two additional cases to the literature and delineated them with Seckel's cases 1 and 2, and his literature cases 1 and 6 (Seckel, 1960) from other bird-headed dwarfs (Man and Russel, 1959; Black, 1961; de la Cruz, 1963), by their developmental, dental, genital, skeletal, and central nervous system anomalies. McKusick *et al* (1967) discussed three sibs with features of low birth weight, small head, prominent beaked nose, mental retardation, sweet disposition, strabismus, sparse hair, and other congenital malformations. In addition, the brain revealed a striking deficiency of convolutions in the areas associated with higher intellectual functions. Since there was a similarity

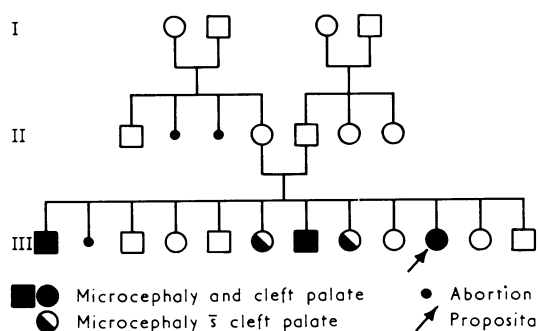


FIG. 3. Family pedigree revealing familial incidence of microcephalic dwarfism with instances of concurrent cleft palate.

to the family Pongidae (chimpanzee) the term pongidoid microencephaly was proposed. An autosomal recessive mode of inheritance was suggested, but was considered separate and distinct from recessively inherited 'true microcephaly' (Komai, Kishimoto, and Ozake, 1955; van den Bosch, 1958/59; Kloepper, Platou, and Hansche, 1964). Warkany (1971) observed a case cited by Horwood in which two children in a family, after several normal pregnancies, delivered with severe intrauterine growth retardation, and bird-headed facies. The fifth pregnancy terminated with the delivery of a still-born infant weighing 920 g. The fetus showed microcephaly and cleft lip and palate. The sixth pregnancy terminated at term with delivery of a female infant weighing 1080 g. There was microcephaly and a high arched palate.

The significance of dental alterations in Seckel's bird-headed dwarfism resides in the defect, hypoplastic enamel, being limited to the primary dentition; in most instances the second primary molar tooth is not affected (Seckel, 1960; Harper *et al*, 1967). The dentition is an excellent structural and temporal indicator of the development of mineralized tissues since its development and eruption from childhood through adolescence provides an indication of the mineralization process that is operative in a complex morphological system during a highly select period of development. The fact that enamel and dentine are not subject to influences of remodeling as is bone provides a permanent measure of those developmental periods. Thus, the ascertainment of highly select dental defects in the dentition of Seckel dwarfism is at least suggestive that some influence, possibly an altered operon unit or regulator gene was temporally altered during the first trimester of pregnancy and more specifically in the third or fourth month of pregnancy. However, more embryonic data and kindreds are necessary

before the establishment of any universal statement regarding the pathogenesis of this syndrome of microcephaly.

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